

ClinVar

NCBI's ClinVar is a freely available submission-driven database for information about genomic variation and its relationship to human health.



ClinVar accepts submissions interpretations of genetic data from:

- clinical genetics testing laboratories
- research groups
- expert panels
- and others



Interpret your data and guide your diagnosis

ncbi.nlm.nih.gov/clinvar

- 1,670+ submitters
- 75+ countries
- 841,000+ variants
- 1,300,000+ submitted records
- [ClinVar Search Video](#)



Contact us at
clinvar@ncbi.nlm.nih.gov



Follow us on Twitter
[@ncbi_clinical](https://twitter.com/ncbi_clinical)



Visit us at ncbi.nlm.nih.gov/clinvar/ to find out more



ClinVar aggregates
**clinical assertions about variants provided
by clinical genetics testing laboratories
and others.**



ClinVar helps clinicians
**interpret genetic test results
and diagnose disorders to
improve patient outcomes.**

What's New

Tailored [notifications](#) for changes in clinical interpretation of variants

Search in ClinVar

[Visit Now](#)



Gene symbols



Diseases and Phenotypes



HGVS expressions



Submitting organization



RS numbers



PubMed ID or other citation



Protein changes

[Download from the FTP Site](#)

Free, current, expert-authored, and peer-reviewed descriptions of heritable disorders

ABOUT GENEREVIEW®

- The busy clinician's #1 source for diagnosis, management, and genetic counseling information
- 780+ chapters with links to disease-specific and general consumer resources
- Includes chapters with a single-gene or -phenotype focus and overviews covering the genetic causes of common conditions
- Educational materials to clarify genetics concepts for patients and general clinicians
- Resources for genetics professionals on genetic testing, including founder variants and direct-to-consumer testing
- New and/or updated chapters published weekly
- Content linked to [ClinVar](#), [MedGen](#), [GTR®](#), [PheGenI](#), [Medical Genetics Summaries](#), and [dbGaP](#)

Search GeneReviews® By:



Disorder



Text



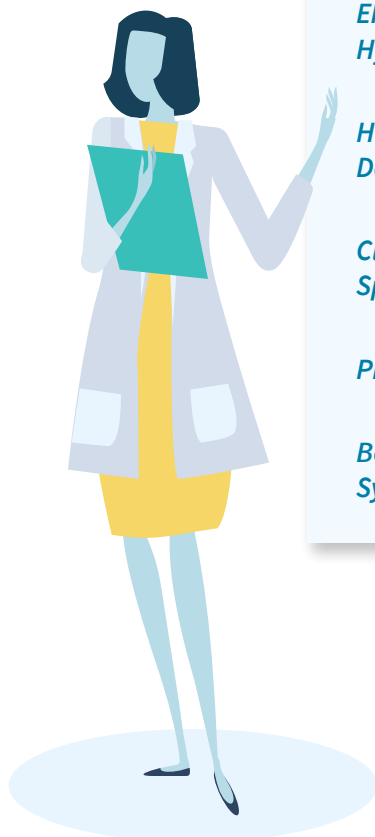
Author



Gene Symbol



Protein Name



GeneReviews® Chapters

*Ehlers-Danlos Syndrome,
Hypermobility Type*

*Hereditary Hearing Loss and
Deafness Overview*

*Cleidocranial Dysplasia
Spectrum Disorder*

Prader-Willi Syndrome

*Beckwith-Wiedemann
Syndrome*

Volunteer to create a GeneReviews® chapter by going to our website and clicking on “For Current/Prospective Authors”

Visit GeneReviews

NIH Genetic Testing Registry (GTR®)

An unbiased, free to participate in, and free to use, international database of clinical and research molecular, cytogenetics and biochemical genetic tests, and supporting information

ABOUT GTR®

- Single gene tests, panels, genomes, and exomes
- 76,000+ tests (incl. 1,600+ tests for somatic targets), 16,000+ conditions, 18,500+ genes, 575+ labs
- 56 BRCA1 single gene tests and 391 multigene panels
- 78 BRCA2 single gene tests and 445 multigene panels

GTR® now includes [molecular and serological tests for microbes](#) that affect human health & disease

- 18 [COVID-19](#) tests
- 15 tests for other viruses, parasites, bacteria

GTR® is a central location for laboratories to provide genetic test information and for clinicians and researchers to search and find genetic tests. GTR® increases transparency in the genetic testing landscape.



Search by

- Test name
- Test services like custom mutation-specific / carrier testing
- Gene, number of genes, or germline vs. somatic
- Analytes / chromosomal regions / proteins
- Lab and staff name, location, or certifications
- Test purpose or specimen type
- Disease or phenotype
- Methodologies

Starting October 2020, register your microbe tests, including:

- molecular tests to detect microbe nucleic acids
- tests to detect microbe-specific antigens
- tests to detect antibodies to a microbe
- microbe panels
- viral load tests to monitor disease progression and guide treatment

LEARN ABOUT GENETIC TESTS AVAILABLE TO YOU

[Visit GTR](#)



Purpose and limitations



Clinical utility



Methodology



Clinical and analytical validity



Lab contacts and credentials, including CLIA and state licenses



AMA CPT® and LOINC codes



Evidence of the test's usefulness



Test ordering information





ACCESS RESOURCES

Bookmark MedGen and use it as the starting point to brush up on genetic phenotypes and navigate to authoritative resources with information about the phenotype. You can quickly be fully prepared for your patient visits!



IMPROVE OUTCOMES

Access practice guidelines from medical and professional societies to provide effective treatment options for your patients. Share consumer resources to educate your patient and family about the diagnosis and prognosis



SEARCH CLINICAL FEATURES

Search aggregated data by clinical feature(s), genes or other attributes, to help inform a differential diagnosis and hone patient research with access to the latest clinical studies, clinical trials, and systematic reviews

RESOURCES

MedGen supports research, diagnosis and treatment of genetic disorders by providing information on:

- Mendelian disorders
- Pharmacogenetic responses
- Complex diseases
- Clinical findings

TOOLS

MedGen's all-in-one platform connects clinicians to leading genetic resources, including:

- PubMed
- GARD
- GeneReviews®
- OMIM



Visit MedGen



MedGen, ClinVar, and GTR®

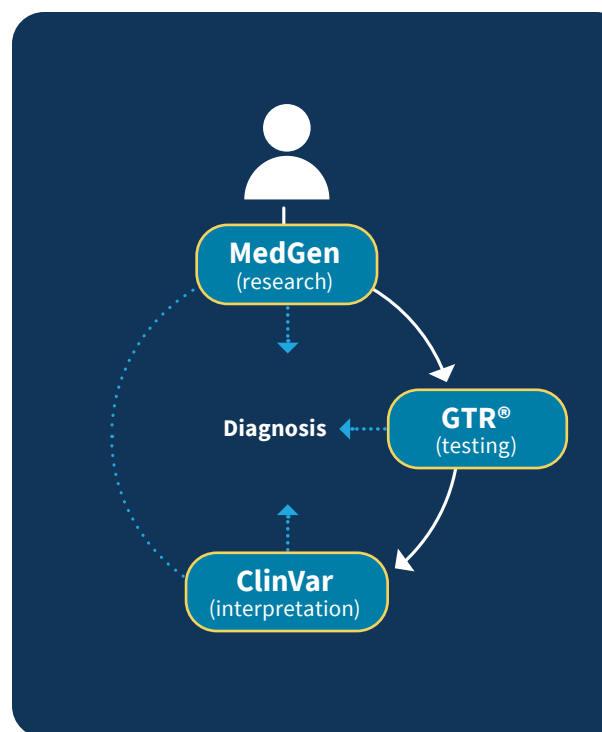


Using NCBI's medical genetics and human variation resources to research, diagnose and treat genetic conditions.

CASE STUDY: SUSPECTED MARFAN SYNDROME

A 9-year-old boy comes into the clinic for a medical release to play soccer. The child presents with myopia, arachnodactyly and has a family history of aortic dissection. These are flags that prompt you to investigate a possible genetic disease.

- 1** Search [MedGen](#) to help develop a differential by patient's clinical features in final diagnosis and learn about a condition, its diagnosis and etiology
- 2** Search the NIH [Genetic Testing Registry \(GTR®\)](#) to find the most appropriate genetic test for your patient – for example a panel that includes all the conditions in your differential diagnosis. Learn about the test's validity and utility, and find the ordering information
- 3** Following testing, compare the test results to interpreted variants in [ClinVar](#) and determine pathogenicity
- 4** Return to [MedGen](#) for practice guidelines from medical and professional societies and the latest research to guide your treatment options. Access consumer resources to help your patient and his family understand his diagnosis, prognosis and available help. If the family is interested, check if there are available studies in [ClinicalTrials.gov](#)



ABOUT OUR RESOURCES

ClinVar has more than 1,300,000 submitted records representing more than 841,000 unique variants from 1670+ submitters.

GTR® has 76,000+ tests for 16,000+ conditions and 18,500+ genes, from 575+ labs. It includes clinical and research molecular, cytogenetic and biochemical genetic tests.

MedGen helps research thousands of genetic phenotypes including Mendelian disorders, complex diseases, clinical features and drug responses. It aggregates information from authoritative resources so from one website you can access most available clinical, consumer and molecular resources.

Share Your Data
with ClinVar

Share Your Data
with GTR



Medical Genetics Summaries (MGS)

Free and growing collection of clinical pharmacogenetics summaries. Each summary reviews one drug, its use and metabolism, the genetic variants that influence how an individual may respond to the drug and aggregates therapeutic guidelines from authoritative sources.

FAST FACTS FOR BUSY CLINICIANS

- Up-to-date, peer-reviewed, actionable pharmacogenetic information
- Access all available therapeutic recommendations based on genotype from the FDA drug label, medical and professional societies, including CPIC, DPWG, ACMG, ASCO
- Introduction gives a clinical context
- Summarizes latest research and evidence for easy consultation at point-of-care
- Synthesizes drug and gene information
- Genetic testing strategies and links to relevant available tests in the NIH Genetic Testing Registry ([GTR®](#))
- Nomenclature table that translates all naming conventions of relevant alleles and links to [ClinVar](#) and [dbSNP](#)



STANDARDIZED FORMAT



Introduction: Summarizes key points and dosage guidelines



Gene(s): Genetic variation that influences drug metabolism



Therapeutic Recommendations based on Genotype excerpted from FDA, CPIC, and other authoritative sources



Drug: including indications, risks and dosage considerations



Genetic Testing: Resources for using and interpreting genetic testing

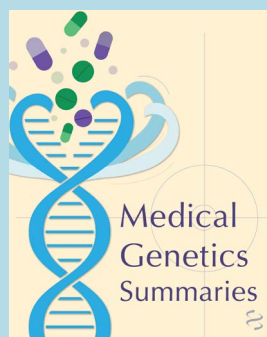


Nomenclature: Allele translation table including star alleles and HGVS

SAMPLE MGS CHAPTERS

[Visit Now](#)

- Codeine
- Tramadol
- Irinotecan
- Tamoxifen
- Pertuzumab
- Flurouracil
- Venlafaxine
- Thioguanine
- Trastuzumab
- Capecitabine
- Mercaptopurine
- And many, many more...



MGS VIDEOS:

[Pharmacogenetics](#)

[Medical Genetics Summaries Overview](#)



Variation Resources



NCBI's variation resources offer human genomic variations, including common and rare SNV, other small-scale variations, large structural variations, and associated frequencies, including ALFA, a new aggregated frequency source based on data from millions of controlled-access research study subjects. Access through the web, APIs, and FTP downloads.

Identify
novel variants

Filter individuals'
genome data

Annotate with
other data such as
genomic features,
Genes & Pubmed
citations

Prioritize variants
likely to impact
biological function

dbSNP

[Visit dbSNP](#)

- Over 2 Billion submissions including data from 1000 Genomes, GnomAD, and others
- 720 Million RS
- Frequency for more than 606 Million RS; including common and rare variants
- Rich annotation reported on RefSeq GRCH37 and GRCH38 assemblies, mRNA, and Protein
- VCF files for assemblies GRCh37 and GRCh38
- Full set of RefSNPs in the JSON format
- [Indexed Search](#)

dbVar

[Visit dbVar](#)

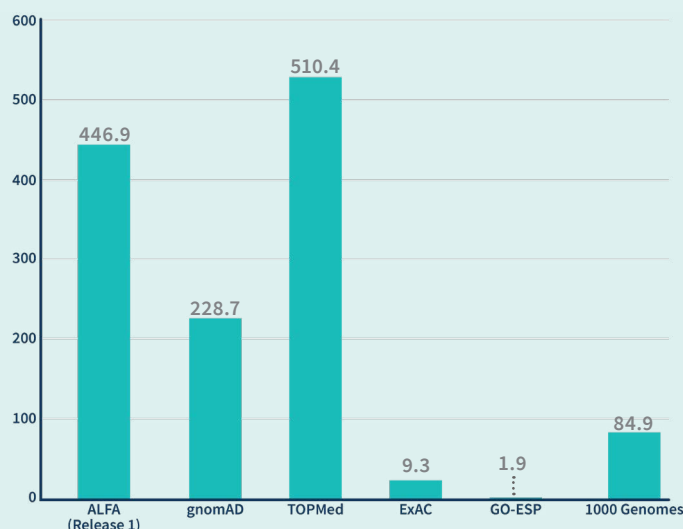
- 193 studies
- Clinically significant SV, Case-Control, and Curated [Datasets](#)
- 6.0 million unique structural variants
- 36.1 million submitted variant calls
- Updated monthly
- Population allele frequency
- Files are available in XML, GVF, VCF, BED, BEDPE, and TSV for assemblies GRCh37 and GRCh38
- [dbVar Tutorials and Datasets](#)
- Access full set of [FTP](#) files

ALFA

[Visit ALFA](#)

- Release 1 (March 2020) included 447M variants from 98K subjects
- Release 2 (October 2020) will include an additional ~100K subjects for a total of ~200K
- Access ALFA data along with other projects including GnomAD, and TOPMed

Variants with frequency data (by project in, million)



Variation Services

Web services for comparing, normalizing, annotating, and inter-converting variations

[Visit Now](#)

Variation Viewer

View, search, and navigate variations in genomic context. Review data or upload your own data

[Visit Now](#)

